



MTM1 gene

myotubularin 1

Normal Function

The *MTM1* gene provides instructions for producing an enzyme called myotubularin. Myotubularin is thought to be involved in the development and maintenance of muscle cells. This enzyme acts as a phosphatase, which means that it removes clusters of oxygen and phosphorus atoms (phosphate groups) from other molecules. Myotubularin removes phosphate groups from two molecules called phosphatidylinositol 3-phosphate and phosphatidylinositol 3,5-biphosphate. These molecules are found within cell membranes and are likely involved in transporting molecules within cells.

Health Conditions Related to Genetic Changes

X-linked myotubular myopathy

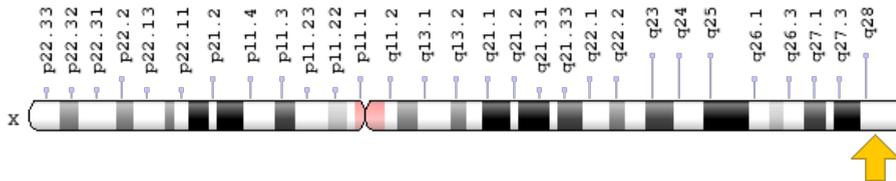
More than 200 mutations in the *MTM1* gene have been found to cause X-linked myotubular myopathy. Some *MTM1* gene mutations change one of the protein building blocks (amino acids) in myotubularin, while other mutations result in an abnormally short, nonfunctional enzyme. The *MTM1* gene mutations that prevent the production of any functional myotubularin tend to result in a more severe disease. Individuals who are mildly affected tend to have an *MTM1* mutation that allows some functional myotubularin to be produced.

Mutations in the *MTM1* gene are thought to disrupt myotubularin's role in muscle cell development and maintenance, causing muscle weakness and other signs and symptoms of X-linked myotubular myopathy.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 150,562,658 to 150,673,143 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CNM
- MTM1_HUMAN
- MTMX
- myotubularin
- XLMTM

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): A Eucaryotic Cell Contains a Large Collection of Protein Kinases and Protein Phosphatases
<https://www.ncbi.nlm.nih.gov/books/NBK26911/#A503>

GeneReviews

- X-Linked Centronuclear Myopathy
<https://www.ncbi.nlm.nih.gov/books/NBK1432>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MTM1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D>

OMIM

- MYOTUBULARIN
<http://omim.org/entry/300415>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MTM1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MTM1%5Bgene%5D>
- HGNC Gene Family: Myotubularins
<http://www.genenames.org/cgi-bin/genefamilies/set/903>
- HGNC Gene Family: Phosphoinositide phosphatases
<http://www.genenames.org/cgi-bin/genefamilies/set/1079>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7448
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4534>
- UniProt
<http://www.uniprot.org/uniprot/Q13496>

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